【演者】

Ype Elgersma

ENCORE expertise center for neurodevelopmental disorders; Department of Neuroscience; Erasmus MC University Medical Center, Rotterdam, The Netherlands

【演題】

Understanding the mechanisms underlying Angelman Syndrome and identifying treatments

【講演要旨】

Intellectual disability (ID) is present in 2% of all children. Recent evidence suggests that in the majority of the cases a genetic mutation is the primary cause of the intellectual disability. Knowledge of the role of mutated gene is an important step in trying to understand the molecular mechanisms leading to ID, and helps us to design therapeutic strategies in which we try to ameliorate the symptoms associated with these disorders.

In this presentation I will in particular focus on our research on Angelman Syndrome. Angelman syndrome (AS) is a severe neurological disorder, affecting 1:20,000. Children with AS present with severe intellectual disability, motor dysfunction and absence of speech. In addition, there is a high comorbidity with epilepsy and behavioral deficits. AS is caused by spontaneous mutations in the *UBE3A* gene. Currently, there are no effective treatments for this devastating disorder. Therefore, the primary aim of our research is to advance our knowledge of the underlying pathophysiology of AS with the ultimate aim to identify promising approaches for improving therapeutic treatment.